A CASE OF MICROCEPHALUS WITH MICROPHTHALMUS

BY

DR. E. PFLUEGER

PROFESSOR OF OPHTHALMOLOGY AT THE UNIVERSITY OF BERNE, SWITZERLAND

Translated by Dr. A. SCHAPRINGER, New York

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(*With plate i.*)

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THE case detailed here is that of Emma Blaser, aged eleven, of Luetzelflueh, Switzerland, who was an inmate of our clinic from November 23 until December 4, 1882. She was seen again in June, 1883, and exhibited to the Medico-Pharmaceutical Society of Berne. The microcephalus was well marked, though only one of a medium degree, and of the variety called trigonocephalus.

The measurements of the skull were made by Professor Flesch, who has had considerable experience in this kind of work, and who was assisted by Dr. Freund. The method employed was that of Virchow and Rieger.

I am indebted to Professor Flesch for the outlines of the head, reproduced in plate i., and for the following data.

" Hair blonde, eyes brown, the same as her mother. Length of body, 133 *cm.*; height to hip, 78.9 *cm.*; length of arm, 57 *cm.* Measurements of skull: greatest length, 158 *mm.*; greatest breadth, 130 *mm.*; horizontal circumference around occipital protuberance and eyebrows, 476 *mm.*; circumference from glabella to occipital protuberance, 290 *mm.*; from glabella to lowest point of occiput between muscles of neck, 328 *mm.*; frontal circumference, above external auditory canals, 299 *mm.*; breadth of skull between zygomatic arches, 100 *mm.*; between openings of audi-

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tory canals, 136 *mm.;* height of face, 109 *mm.;* of nose, 59 *mm.;* breadth of face between rami of lower jaw, 86 *mm.;* length of right half of lower jaw, 73 *mm.;* of left, 71 *mm.*

"The two lateral halves of the skull are asymmetrical, the left being less developed anteriorly, the right less developed posteriorly. The line of circumference shows a marked indentation in the temporal region of the right side, and consequently the greatest breadth is found far back. The most striking feature of the head is the narrowness of its frontal portion, which constitutes it a trigonocephalus. The general measurements of the head are not excessively small, except those of the frontal segment, the diminutive proportions of which make it evident that there must be a deficient development of the brain."

The microphthalmus, like the microcephalus, while not of high degree, was well marked and unmistakable. The corneæ were not circular, but transversely elliptical, the vertical diameter of each being 9 *mm.*, the horizontal 10 *mm.*

There were two symptoms present which rendered the examination of the eyes difficult and incomplete—very violent horizontal nystagmus and complete absence of central fixation. The little girl habitually held her head inclined to the left, often to such an extent that the bridge of her nose was parallel with the line of her shoulder, her eyes being rolled to the extreme right, and at the same time considerably either above or below the horizontal meridian.

The acuteness of vision could not be determined in either eye, owing partly to the great imperfection of the visual power, and partly to the deficient development of her intellect. It was evident, however, that she used only her right eye for fixation, fixing not with the macula lutea of this eye, but with a portion of the retina, situated a considerable distance to the outer side of the macula, and either above or below the horizontal meridian. The refraction of the right eye, as determined by keratoscopy, proved to be slightly myopic (1 or 2 D.).

Both corneæ were perfectly transparent. The iris of either eye was normal in every respect, except as to size. There was no coloboma, the pupils being normal in shape, size, and position. Atropia caused only incomplete dilatation of the pupils.

The lens of the right eye was completely transparent. That of the left showed partial posterior cortical cataract, which will be described further on, and on account of which I performed a

small pear-shaped iridectomy on the nasal side. Although the optical conditions of the eye were greatly improved by this operation, her visual power did not seem to increase.

In the background of both eyes were numerous irregular yellowish-white atrophic patches, varying in size from a mere dot to that of the optic disc, apparently the sequelæ of intra-uterine retino-chorioiditis. There were also numerous oblong collections of dark pigment, the length of some being equal to five or six times the diameter of the disc. The most remarkable feature, however, were large greenish-blue spaces of oblong form, showing a reflex like that of mother-of-pearl.

In both eyes there was a coloboma of the sheath of the optic nerve and of the choroid on the temporal side of the optic disc. In the right eye the shape of the disc was a transverse oval. It was well defined by the scleral ring above, below, and on the nasal side. On the temporal side the vaginal ring was absent, and its place was taken by translucent nerve-substance. Here the boundary line of the disc was less distinct than normal, and showed indentation which made the disc appear slightly heart-shaped.

The choroid receded from the lower periphery of the disc, less on the nasal than on the temporal side ; hence the appearance of a spherical triangle with an acute angle at the nasal portion of the disc. Passing from the nasal to the temporal side of the disc, the distance of its periphery from the choroidal margin increased, and the choroid, instead of rising on the temporal side in correspondence to the outline of the disc, kept its direction outward and downward, being sharply defined for some distance only. At the upper periphery of the optic disc the choroidal border receded from the disc, as below, continuing directly outward and upward across the fundus, instead of lining the temporal border of the papilla. The two sides of the choroidal coloboma formed an angle of 50° to 60°, which was bisected by the horizontal meridian of theglobe.

The coloboma did not show the bright yellowish-white color of the bare sclera throughout, but only in areas which were divided by accumulations of pigment distributed with a certain degree of regularity. The pigment lay in vertical streaks slightly curved, the convexity of the curves looking toward the temple. This regularity was most striking in the immediate vicinity of the disc, so that the lower crescent appeared to be composed of a number

of alternately black and yellow vertical arches. This regularity was less marked, however, on the temporal side.

At a distance of from 6–7 P. D., and situated at its outer side, there was a curved oblong band running vertically, its length equal to 6–8 P. D., and its breadth to about 1 P. D., which showed a brilliant greenish-blue reflex. On the outer side of this band ran a streak of very black pigment, having at its broadest portion a breadth equal to $\frac{1}{4}$ P. D. On the outer side of this streak the choroid showed the same appearance as in the rest of the background, being studded with choroidal exudations, atrophic patches, and numerous accumulations of pigment.

The arrangement of the retinal vessels was strikingly abnormal. The main vessels did not run upward and downward, but they all ran outward and formed an angle of 45° to 50°. One small vessel ran upward and inward upon the papilla, and another small one downward and inward. Upon reaching the border of the disc both suddenly turned and took an outward course, only to change it again after running a short distance by returning to the disc and traversing it from without inward. The two vessels then crossed each other, presenting the appearance of a bow-knot.

The fundus of the left eye presented changes more pronounced and similar, with the following exceptions: There was also a coloboma of the sheath of the optic nerve and of the choroid. The sides of the choroidal coloboma, however, did not diverge, but ran parallel to each other. They could not be traced further than the equator, on account of the cortical opacity of the lens. The optic disc was not quite so small, but it was more elliptical, its vertical diameter being somewhat smaller than that of the right disc. The color of the coloboma was darker, with an occasional greenish shimmer, and extensive and irregular accumulations of pigment. The portions of the choroid immediately adjoining the coloboma showed little change, but toward the periphery it had undergone extensive changes. Here, as well as in the other eye, the atrophic patches were greatly outnumbered by the proliferations of pigment. There was also an extensive area of a greenish-blue color, resembling the tapetum of animals. Unfortunately, it was not possible to thoroughly study all the details.

The retinal vessels were parallel and ran directly outward, following the direction of the sides of the choroidal coloboma. There were no retinal vessels running in any other direction.

The calibre of the retinal vessels seemed to be smaller than

normal, more markedly so in the left eye than in the right. The arteries and veins could not be distinguished from each other.

The color and translucency of the right optic disc was about normal, but the left was as opaque as in cases of advanced retino-choroidal atrophy.

The opacity of the left lens mentioned above was situated mainly in the posterior cortical substance. It began at the posterior pole, and was of such shape and size that it just obstructed the pupillary area when looked at from in front, before the iridectomy was performed. From the posterior pole the opacity extended outward as far as the equator, forming an isosceles triangle with the base at the equator. The rounded angle at the posterior pole was about 40°. The opacity did not cease at the equator, but extended for the distance of 1.5 to 2 *mm.* into the anterior cortical layer. There was evidently a causal relation between this opacity of the lens and the choroidal coloboma, as evidenced by the correspondence of their location.

It does not seem to be difficult to find an explanation for this anomaly of development. Manz, noticing the frequent coincidence of coloboma of the uvea and microphthalmus, suggested that microphthalmus might be caused by intra-uterine chorioiditis. This supposition was confirmed by Deutschmann,[1] who had the opportunity of making an autopsy of such a case. The present one also tends to corroborate the hypothesis of Manz.

In his review of Deutschmann's paper in *Nagel's Jahresbericht* Manz says: "The supposition appears to be very plausible that intra-uterine sclero-chorio-retinitis will cause incomplete closure of the fœtal fissure, or will cause it to re-open if already closed. This hypothesis will certainly be generally accepted if corroborated by the anatomical examination of a few more cases of coloboma bulbi. One thing, however, still remains unexplained, namely the fact that the inflammatory changes are confined to the neighborhood of the ocular fissure."

Our case evidently serves to fill a gap here, as in it the choroidal changes, though most striking in the neighborhood of the coloboma, were well marked throughout the whole fundus.

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[1] *Zehender's klin. Monatsbl.*, Bd. xix., p. 112.

The course of the retinal vessels was very peculiar and such as I had never before observed in the human being. It can be explained by assuming that the cicatricial contraction of the choroidal coloboma caused traction upon the retina, dragging its upper portion downward and the lower upward. The indentation of the outer periphery of the right optic disc can also be explained by the cicatricial contraction of the sides of the coloboma. This theory seems to be corroborated by the concentric arrangement of the pigment deposits in the fundus of the right eye. The elliptic form of the cornea was another feature for which the theory of cicatricial contraction affords a natural explanation. In our case, where the direction of this contraction was vertical, the greater diameter of the cornea was horizontal, whilst in the case published by Deutschmann, in which the direction of the coloboma was downward, the lesser axis of the cornea was horizontal.

The outward direction of the coloboma in this case was unusual, the coloboma being usually directed downwards. This unusual feature can be explained by the hypothesis that the fœtal cleft is first directed outward, and that a gradual rotation of the globe changes this direction downwards, and that in the present case the globe for some reason or other had not performed this rotation.

Frau A. Blaser, æt. forty-four, the girl's mother, showed an abnormal condition of the skull as well as of the eye similar to that exhibited by her daughter only somewhat less marked. The greatest horizontal circumference of the skull was 530 *mm.*, the left half being larger by 5 *mm.* than the right. Circumference around glabella and occipital protuberance 284 *mm.*, over gabella and muscles of the neck 310 *mm.*, arch over auditory orifices 360 *mm.*, length about 170 *mm.*, breadth about 137 *mm.*

Both corneæ were somewhat smaller than normal and slightly elliptical, the vertical diameter being 10 *mm.*, the horizontal 11 *mm.* No coloboma. Choroidal ring at outer periphery of optic disc very dark.

Frau Blaser has another daughter who is said to have perfectly normal eyes. She has had a third child, which died, two hours after birth, from hydrocephalus.

Frau St., æt. thirty-five, a sister of Frau Blaser, has a normal skull, but a slight microphthalmus, the diameter of either cornea being only 10 *mm.*

O. s. M. 0. 75 and As. m. 0.5, axis vertical, V = 0.5
O. d. M. 1 and As. m. 0.5, axis vertical, V = 0.75

Ophthalmoscopic appearances normal.

Frau Blaser's father is also said to have abnormal eyes, but he could not be prevailed upon to present himself for examination.

Explanation of Plate No I.

Projection of shape of head according to Rieger and Virchow (Sitzungsber. d. phys.-med. Ges. in Würzburg, 1882, p. 96).

————Sagittal Arch (Glabella and Occipital Protuberance).

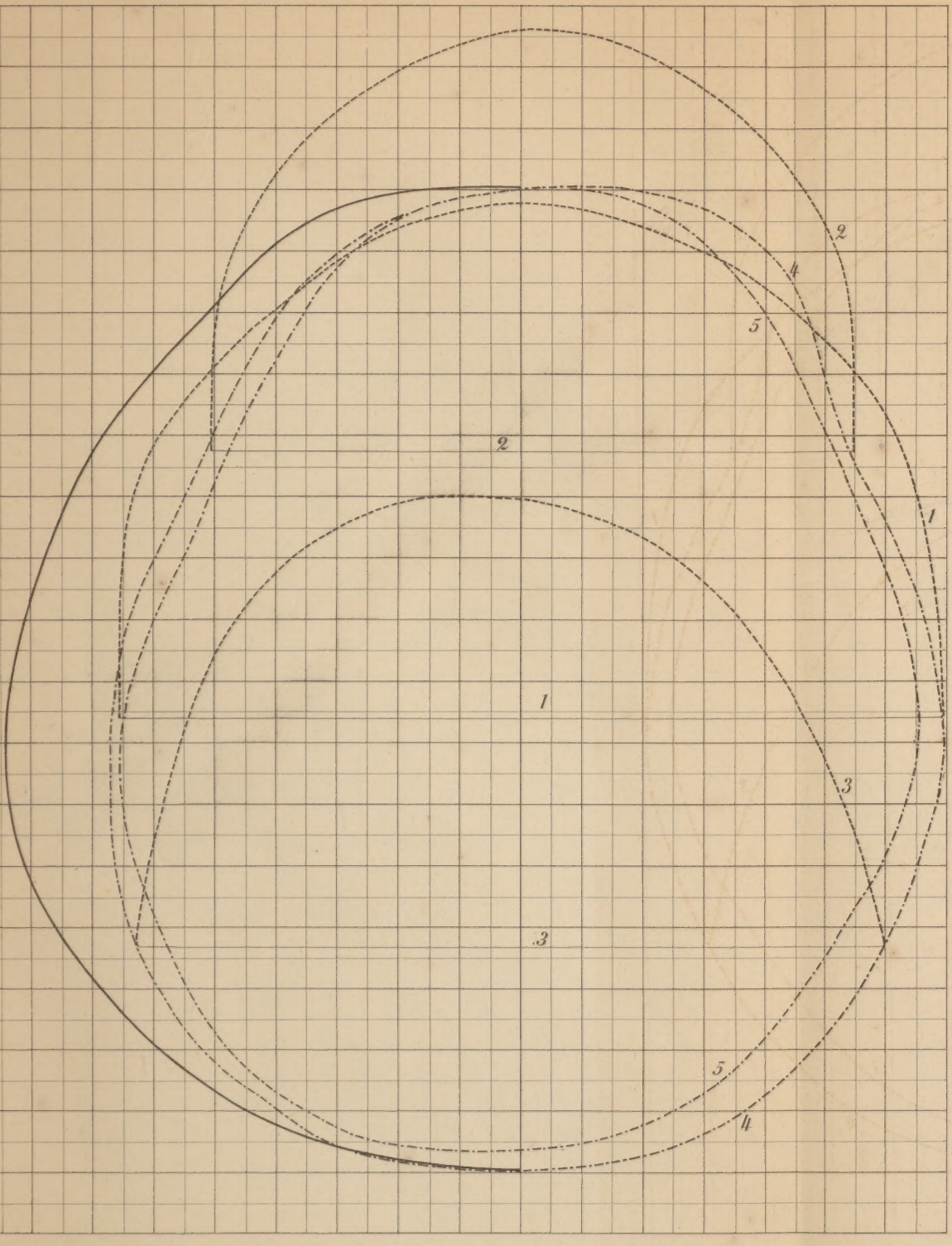
— — — — Frontal Arches :

1. Breadth at auditory orifices.
2. Arch between auditory orifice and glabella.
3. Occipital arch, between auditory orifice and occipital protuberance.

These three arches stand vertically upon the horizontal plane.

—·—·—·—·Horizontal Planes :

4. Through occipital protuberance and glabella.
5. A parallel plane two *cm.* higher than the preceding.



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